

Leukaemia Section

Mini Review

t(9;14)(q34;q32)

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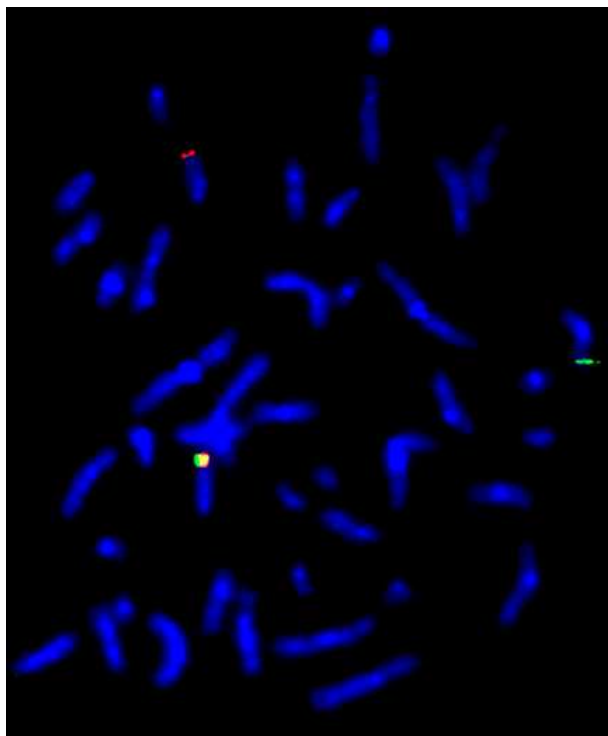
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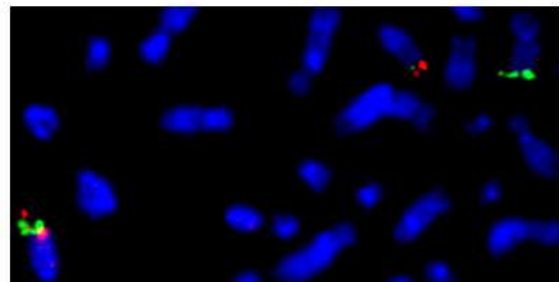
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Identity



t(9;14)(q34;q32):
Top left: IGH3' red, IGH5' green,
right: same metaphase in G-banding
Bottom, another metaphase



t(9;14)(q34;q32) G-banding and FISH - Courtesy Melanie Zenger and Claudia Haferlach.

Clinics and pathology

Disease

T cell acute lymphoblastic leukemia (T-ALL).

Epidemiology

Only 1 case to date: a 16 yr old female patient.

Cytology

High leukocytosis with 99% blasts with the phenotype of cortical thymocytes.

Prognosis

Yet unknown; the patient was in complete remission at 15 mths+.

Cytogenetics

Cytogenetics morphological

Cryptic translocation: the karyotype appeared normal.

Cytogenetics molecular

FISH with 5' ABL1 probe (RP11-57C19) and 3' ABL1 probe (RP11-83J21) resulted in a split signal

Probes

RP11-57C19 and RP11-83J21 (BACPAC Resources, Oakland, CA).

Additional anomalies

Patient had also hemizygous deletion of CDKN2A and ectopic expression of TLX1.

Genes involved and proteins

ABL1

Location

9q34

Protein

Tyrosine kinase.

EML1

Location

14q32

Note

Gene was mapped within Usher syndrome type 1a locus.

Protein

Protein is very similar to the echinoderm microtubule-associated protein.

Result of the chromosomal anomaly

Hybrid gene

Description

5' EML1 - 3' ABL1, in frame fusion between exon 17 of EML1 and exon 2 of ABL1.

Detection

RT-PCR using the primers 5'- cactcactgggaggtggttt and 5'- acaccattccccattgtgattat.

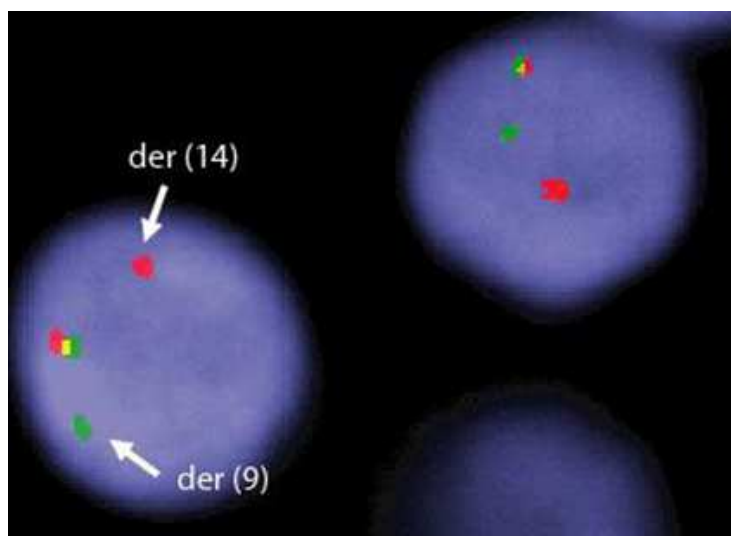
Fusion protein

Description

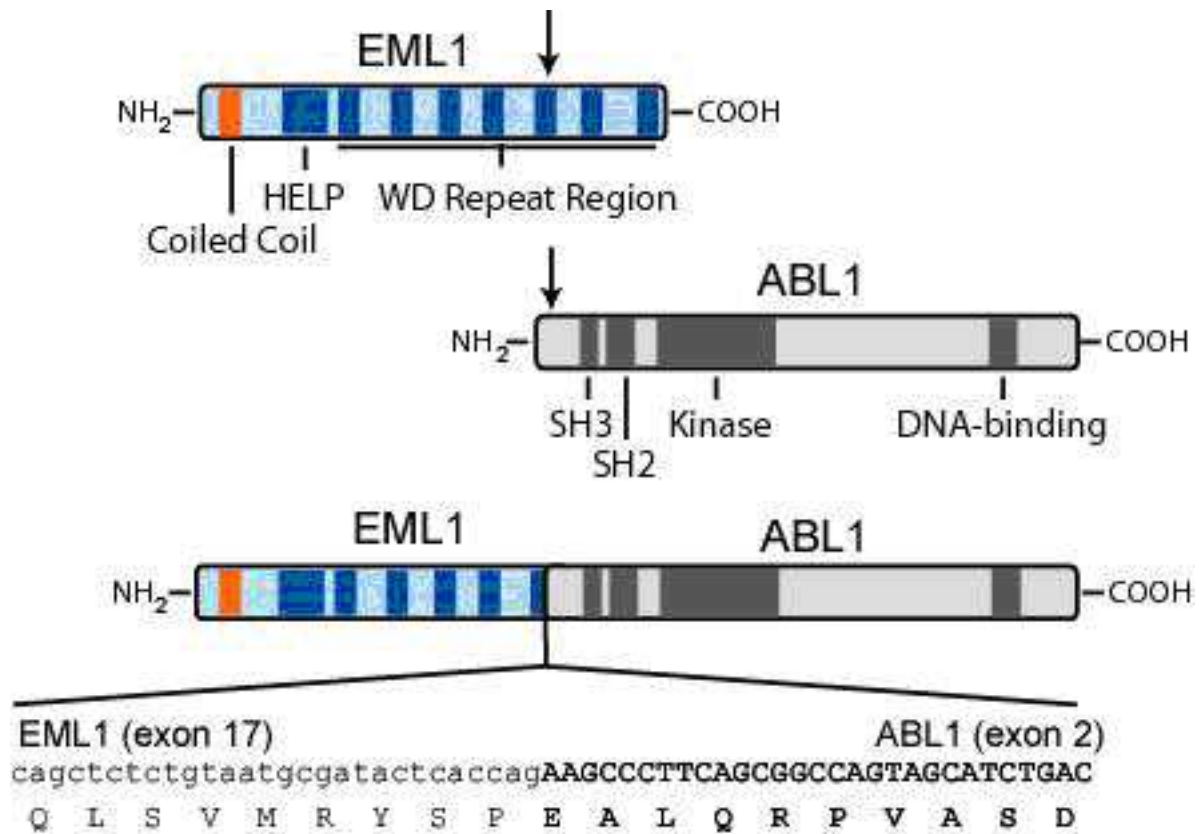
190 kDa NH2 EML1 - COOH ABL1 fusion protein, contains the coiled-coil domain of EML1 and the kinase domain of ABL1.

Oncogenesis

Constitutive EML1-ABL1 tyrosine kinase activity causing deregulation of cellular survival and proliferation pathways.



FISH with 5' ABL1 (green signal) and 3' ABL1 (red signal) probes on metaphase cells of the T-ALL patient with the cryptic t(9;14)(q34;q32). The translocation causes separation of the 2 probes with the 5' ABL1 probe hybridizing to der(9) and the 3' ABL1 probe hybridizing to der(14) - Kim De Keersmaecker.



Schematic representation of EML1 and ABL1 proteins. The EML1-ABL1 fusion protein generated after t(9;14)(q34;q32) is represented beneath. The sequence of the in-frame fusion between exon 17 of EML1 and exon 2 of ABL1 is indicated at the bottom, translation of the nucleotide sequence is shown beneath - Kim De Keersmaecker.

References

De Keersmaecker K, Graux C, Otero MD, Mentens N, Somers R, Maertens J, Wlodarska I, Vandenberghe P, Hagemijer A, Marynen P, Cools J. Fusion of EML1 to ABL1 in T-cell acute lymphoblastic leukemia with cryptic t(9;14)(q34;q32). *Blood*. 2005 Jun 15;105(12):4849-52

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